

CLAIMS

What is claimed is:

1. An isolated nucleic acid molecule encoding an ABCA6 transporter or a functional fragment thereof, wherein said nucleic acid molecule is selected from the group consisting of:
 - (a) a nucleic acid molecule comprising a nucleotide sequence which is at least 90% identical to a portion of, or the full length of, the nucleotide sequence of SEQ ID NO: 1 or 3;
 - (b) a nucleic acid molecule which encodes a polypeptide comprising an amino acid sequence at least about 90% similar to a portion of, or the full length of, the amino acid sequence of SEQ ID NO: 2; and
 - (c) a nucleic acid molecule comprising a nucleotide sequence which hybridizes to the nucleotide sequence of SEQ ID NO: 1 or 3 under stringent conditions.
2. The isolated nucleic acid molecule of claim 1 comprising the nucleotide sequence set forth in SEQ ID NO: 1 or 3.
3. The isolated nucleic acid molecule of claim 1 which encodes a polypeptide comprising the amino acid sequence set forth in SEQ ID NO: 2.
4. The isolated nucleic acid molecule of claim 1 which encodes a polypeptide comprising a naturally occurring allelic variant of the amino acid sequence set forth in SEQ ID NO: 2, wherein said allelic variant binds to an antibody that selectively binds to the polypeptide of SEQ ID NO: 2.
5. An oligonucleotide primer comprising at least 12 contiguous nucleotides of SEQ ID NO: 1 or 3, or a complement thereof from a region specific to ABCA6 transporters.

6. An isolated nucleic acid molecule comprising a nucleotide sequence which is complementary to the nucleotide sequence of the nucleic acid molecule of claim 1.

7. An isolated nucleic acid molecule comprising the nucleic acid molecule of claim 1 and a nucleotide sequence encoding a heterologous polypeptide.

8. A vector comprising the nucleic acid molecule of claim 1.

9. The vector of claim 8, which is an expression vector.

10. A host cell transfected with the expression vector of claim 9.

11. A method of producing a polypeptide comprising culturing the host cell of claim 10 in an appropriate culture medium to, thereby, produce the polypeptide.

12. An isolated polypeptide selected from the group consisting of:

(a) a polypeptide comprising the amino acid sequence of SEQ ID NO: 2 or a functional fragment thereof;

(b) a polypeptide comprising a naturally occurring allelic variant of a polypeptide comprising the amino acid sequence of SEQ ID NO: 2, wherein said allelic variant binds to an antibody that selectively binds to the polypeptide of SEQ ID NO: 2;

(c) a polypeptide comprising a functional fragment of the allelic variant of (b);

(d) a polypeptide comprising an amino acid sequence which is at least 90% similar to the amino acid sequence of SEQ ID NO: 2, wherein said amino acid sequence is an ABCA6 transporter; and

(e) a polypeptide comprising a functional fragment of the amino acid sequence of (d).

13. The polypeptide of claim 12, further comprising at least one heterologous amino acid sequence at the amino- or the carboxyl- terminus of said polypeptide.

14. An antibody which selectively binds to a polypeptide of claim 12.

15. A method for detecting the presence of a polypeptide of claim 12 in a biological sample comprising:

- (a) contacting the sample with a compound which selectively binds to the polypeptide; and
- (b) detecting the presence of a complex between said compound and said polypeptide.

16. The method of claim 15, wherein the compound which binds to the polypeptide is an antibody.

17. A kit comprising a compound which selectively binds to a polypeptide of claim 12 and instructions for use.

18. A method for detecting the presence of a nucleic acid molecule of claim 1 in a biological sample comprising:

- (a) contacting said sample with a nucleic acid probe or primer which selectively hybridizes to the nucleic acid molecule; and
- (b) detecting the presence of a complex of said nucleic acid molecule and said probe or primer.

19. The method of claim 18, wherein the sample comprises mRNA molecules and is contacted with a nucleic acid probe.

20. A kit comprising a compound which selectively hybridizes to a nucleic acid molecule of claim 1 and instructions for use.

21. A method for identifying a compound which binds to a polypeptide of claim 12 comprising:

- (a) contacting said polypeptide, or a cell expressing said polypeptide with a test compound; and
- (b) determining whether the polypeptide binds to the test compound.

22. The method of claim 21, wherein the binding of the test compound to the polypeptide is detected by a method selected from the group consisting of:

- (a) direct detection of binding;
- (b) detection of binding using a competitive binding assay; and
- (c) detection of binding using an assay for ABCA6 transporter activity.

23. A method for modulating the activity of a polypeptide of claim 12 comprising contacting said polypeptide or a cell expressing said polypeptide with a compound which binds to the polypeptide in a sufficient concentration to modulate the activity of the polypeptide.

24. A method for identifying a compound which modulates the activity of a polypeptide of claim 12 comprising:

- (a) contacting a polypeptide of claim 12 with a test compound; and
- (b) determining the effect of the test compound on the activity of the polypeptide to thereby identify a compound which modulates the activity of the polypeptide.

25. A method for detecting an allelic variation of the nucleic acid of SEQ ID NO: 1 or an orthologue thereof in a biological sample, comprising:

(a) obtaining from said sample a polynucleotide that hybridizes to the nucleic acid of SEQ ID NO:1 or the orthologue thereof; and

(b) determining whether said polynucleotide is identical to a portion, or the full length, of SEQ ID NO: 1 or the orthologue thereof.

26. A composition comprising a pharmaceutically effective amount of the nucleic acid molecule of SEQ ID NO: 1 or a functional fragment thereof and a pharmaceutically acceptable carrier.

27. A composition comprising a pharmaceutically effective amount of an antisense oligonucleotide capable of specifically hybridizing to a portion, or the full length, of SEQ ID NO: 1 and a pharmaceutical acceptable carrier.

28. A transgenic knockout mouse whose genome comprises a homozygous disruption in its endogenous ABCA6 gene, wherein said homozygous disruption prevents the expression of a functional ABCA6 protein, and wherein said homozygous disruption results in said transgenic knockout mouse being sterile.